■ Texas Department of Health ■

MONITOR

■ Bureau of Epidemiology ■

TEXAS BIRTH DEFECTS MONITORING DIVISION

VOLUME 7, NUMBER 2, DECEMBER 2001

FROM THE DIRECTOR

Ten Years After ...

It has been 10 years since three infants with anencephaly were delivered in a 36-hour period in one facility in Brownsville, Texas. Astute clinicians recognized that this was excessive for this period and facility, and they alerted the Texas Department of Health (TDH) of this cluster. Over the next several years, TDH, in cooperation with local officials and providers and the Centers for Disease Control and Prevention (CDC), conducted a thorough epidemiologic investigation of neural tube defects in Cameron County (which includes Brownsville) and Hidalgo County, the two most southeastern Texas counties that border Mexico. Compared with the United States, high rates of neural tube defects, particularly anencephaly, were confirmed for the area, especially Cameron County in 1991. The investigation underscored a general lack of background data on birth defects in Texas.

In response to this cluster and the need for better data, and in recognition of the enormous resources routinely put forth by TDH in the investigation of birth defect clusters, the Texas State Legislature passed the Texas Birth Defects Act in 1993. Out of this statute, the Texas Birth Defects Monitoring Division (TBDMD) was created to actively identify children born with birth defects.

At about the same time, the Texas Neural Tube Defects Project (TNTDP) was initiated by TDH and collaborators. This seven-year study, funded in large part by the CDC, focused on surveillance, research and recurrence prevention in Texas counties along the border with Mexico.

In the intervening years, much has been accomplished to ensure that Texans have access to vital information about birth defects prevalence and trends in the state. Progress has also been made in research and

WHAT'S INSIDE

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intervention studies designed to prevent further birth defects. Below is a summary of other accomplishments and milestones directly and indirectly related to the Brownsville NTD Cluster:

- Creation of Birth Defects Registry, expanding by 1999 to cover all counties in the state (subsequently curtailed due to funding concerns).
- Establishment of a standard protocol for investigating reports of high rates of NTDs and other birth defects. Since 1994, at least fifteen NTD clusters throughout the state have been investigated by TBDMD staff. (Elevated rates have continued to recur periodically in Webb and Cameron counties.)
- Initiation of a statewide NTD Recurrence Prevention Project to carry on the TNTDP intervention component, currently being piloted on the border and West Texas.
- Funding and creation of the Texas Birth Defects Research Center (TBDRC), which has resulted in collaboration on a number of border or NTD studies.
- Launch of the "Texas-Mexico Border States Birth Defects Project," which includes an intervention pilot planned for Nuevo Laredo, Mexico, in collaboration with the City of Laredo Health Department, TBDRC, TBDMD Region 11, Mexican Health officials, TDH Border Health, and PAHO.
- A "Scientific and Community Forum on NTDs in South Texas" was held in Brownsville in April 1997, followed by a statewide birth defects conference in January 2000 (and another upcoming in March 2002).
- Creation of the Texas Folic Acid Council with funding from the March of Dimes, enabling a statewide folic acid and NTD prevention campaign.

It has been estimated that TDH has spent nearly $4 million on NTD surveillance, research, and prevention since elevated rates in some Texas counties were first identified.

As tragic as any cluster of birth defects is for the families involved, Texans can be proud of the public health response to the babies born with anencephaly in Brownsville 10 years ago.

Regional Bulletin

Beginning with births occurring in 2001, the Texas Birth Defects Registry will no longer collect data on birth defects in Public Health Region 4 (northeast Texas), and will have limited surveillance in Region 1 (the Panhandle area). Funding changes at the Texas Department of Health have necessitated scaling back statewide operations. Numerous other administrative adjustments were made to meet the current fiscal situation.

Data will be available for births occurring in 1999 and 2000 in these regions.

From the Registry

Pregnancy outcome patterns for various birth defects

See Special Report insert.

Research in Texas

Effects of Hyperinsulinemia and Obesity on Risk of Neural Tube Defects among Mexican Americans

Authors: Kate A. Hendricks, Olga M. Nuno, Lucina Suarez, Russell Larsen

This is an excerpt from an article published in the November 2001 issue (Volume 12, Number 6) of Epidemiology (Copyright 2001 by Lippincott Williams & Wilkins, Inc.). Investigators work for the Texas Department of Health. This work was done by the Texas Neural Tube Defects Project, supported in part through the Texas Birth Defects Research Center.

Although both maternal obesity and diabetes mellitus increase the risk for neural tube defects, it is unknown whether they are independent risk factors or manifestations of an underlying prediabetic state such as hyperinsulinemia. We investigated whether hyperinsulinemia was a risk factor for neural tube defects independent of obesity and hyperglycemia in Mexican American women. We identified case- and control-women from residents delivering or terminating pregnancies in hospitals or birthing centers in any of the 14 Texas-Mexico border counties during 1995-2000. Case-women had a pregnancy affected by anencephaly, spina bifida, or encephalocele; randomly selected control-women had normal births, frequency matched by year and birth facility. Questionnaire and laboratory values obtained 5-6 weeks postpartum were available for 149 case- and 178 control-women. Both hyperinsulinemia and obesity were related to increased neural tube defect risk [odds ratio (OR) = 1.91, 95% confidence interval (CI) = 1.21-3.01 and OR = 1.73, 95% CI = 1.03-2.92, respectively]. Adjustment for obesity only slightly reduced the effect of hyperinsulinemia [OR = 1.75, 95% CI = 1.09-2.82]. Alternatively, a modest effect remained for obesity after adjustment for hyperinsulinemia [OR = 1.45, 95% CI = 0.84-2.51]. Hyperinsulinemia is a strong risk factor for neural tube defects and may be the driving force for the observed risk in obese women.

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PREVENTION NOTES

Lower folic acid supplementation rates in Texas related to socioeconomic status, but not cost

Amy Case, Mark Canfield, Bodhini Jasayuria, Ken Condon

The following is a summary of a poster presented at the 7th Annual Maternal Child Epidemiology Conference in December 2001, in Clearwater Beach, Florida.

OBJECTIVE: To evaluate the extent of folic acid awareness and supplementation among Texas women ages 18-44.

METHODS: We used data from the Behavioral Risk Factor Surveillance System (BRFSS) in Texas in 1999 and 2000 combined, using the Centers for Disease Control and Prevention's (CDC) national optional module on folic acid/vitamins (n=1421), plus two questions specific to Texas (n=1104).

Women ages 18-44 were asked five questions in standard optional folic acid module:

- Are you currently taking any vitamin pills or supplements?
- Are any of these a multivitamin?
- Do any of the vitamin pills you take contain folic acid?
- How often do you take this vitamin pill or supplement?
- Some health experts recommend that women take 400 mcg of folic acid daily. Why?

In addition, two Texas-specific customized questions were asked:

- Has a doctor, nurse or health care provider ever advised you to take vitamins or supplements?
- What is the main reason you do not take vitamin pills or supplements?

Although there is a significant association between household income and whether or not the woman takes a folic acid supplement daily, only 8.1% of those who do not take any supplements indicate that cost was the major barrier (Can t Afford).

Summary

FOLIC ACID KNOWLEDGE AND BEHAVIOR: As with several preventive health behaviors measured by the BRFSS (Pap smears, regular exercise, smoke detectors, dental care), folic acid supplementation is strongly associated with socioeconomic status.

IMPLICATIONS FOR FOLIC ACID EDUCATION: Nearly one-half of the reasons given for NOT taking folic acid reflect a need for consistent reminders (not a habit, forget, lack of knowledge, no reason), whereas less than 10% indicated that cost was a barrier. This may indicate that social marketing campaigns which include media reminders, promotional materials such as magnets and pill boxes could be more effective than simply providing free or lower-cost vitamins.

For more information, contact Amy Case at 512-458-7232, amy.case@tdh.state.tx.us.

Table 1: Percentage of women of childbearing age from the Texas BRFSS who reported taking a folic-acid containing supplement, by selected sociodemographic characteristics - 1999 & 2000

Characteristics	Used a folic acid-containing vitamin supplement				Did not use a folic acid-containing vitamin supplement	
	Daily		Less than daily			
	Sample Size	%	Sample Size	%	Sample Size	%
Age group (yrs)						
18-24	88	**35.5**	14	**4.9**	151	**59.5**
25-34	215	**36.5**	47	**8.0**	296	**55.5**
35-44	253	**40.6**	54	**8.4**	296	**51.1**
Education						
Less than high school	56	**22.0**	13	**4.8**	183	**73.2**
High school	303	**39.4**	64	**7.7**	387	**52.9**
College or above	199	**47.6**	38	**9.0**	176	**43.4**
Annual Household Income						
<$25,000	129	**24.9**	33	**6.6**	298	**68.5**
$25,000-$49,999	204	**43.9**	33	**6.7**	231	**49.4**
$50,000+	182	**48.3**	39	**9.8**	148	**41.9**
Martial Status						
Married	337	**41.2**	68	**7.7**	393	**51.2**
Unmarried	139	**32.5**	31	**7.2**	227	**60.3**
Race/Ethnicity						
Non-Hispanic White	353	**45.4**	65	**8.0**	345	**46.6**
Black	52	**32.0**	14	**6.1**	92	**61.9**
Hispanic	127	**27.7**	30	**6.4**	273	**65.8**
Other	23	**47.1**	6	**15.1**	24	**37.8**
Total	559	**38.0**	115	**7.4**	747	**54.6**
Knows that Folic Acid Prevents Birth Defects						
Yes	252	**46.6**	60	**10.7**	226	**42.7**
No	198	**33.4**	43	**6.5**	300	**60.0**
Provider Recommended						
Yes	241	**50.2**	47	**8.6**	207	**41.1**
No	171	**30.2**	39	**7.6**	292	**62.1**

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The authors gratefully acknowledge the contributions of Shirley Baker and Jason Vendel, Public Policy Research Institute, Texas A&M University.

CALENDAR

MARCH 7 - 8, 2002: The Texas Birth-Defects Conference 2002 Radisson-Plaza Hotel, Ft. Worth (See box, right.)

APRIL 21-22, 2002: Human Teratogens, Massachusetts General Hospital, Boston, MA. Phone, 617-384-8600.

SEPTEMBER 30 - OCTOBER 3, 2002: Statewide Community Resource Coordination Groups (CRCGs) of Texas Conference 2002, San Antonio. Texas Health and Human Services Commission, (512) 424-6500.

TEXAS BIRTH DEFECTS CONFERENCE 2002

The only statewide conference focused entirely on birth defects - research, prevention and treatment.

Fort Worth, March 7th and 8th at the Radisson Plaza in Downtown Fort Worth, 818 N. Main Street.

Texas professionals in the fields of maternal/child health nursing, genetic counseling, perinatology, epidemiology, and health education are encouraged to attend. Family members are welcome. Conference offers Continuing Education Units for nurses, physicians, social worker, genetic counselors, and mental health professionals.

Register online at www.tdh.state.tx.us/tbdmd/conf_page.html For more information, please contact the Texas Birth Defects Monitoring Division, 512-458-7232 or amy.case@tdh.state.tx.us

ANNOUNCEMENTS

ON AUGUST 3 SENATOR EDWARD KENNEDY AND SENATOR ORRIN HATCH INTRODUCED THE RARE DISEASES ACT OF 2001 (S. 1379). This bill was placed on the Senate Legislative Calendar on December 18, 2001, and amends the Pubic Health Service Act to establish an Office of Rare Diseases at the National Institutes of Health and provides for rare disease regional centers of excellence, which will include research and educational duties. Appropriations of $20 million are authorized for fiscal year 2002 for the establishment of these centers. The Act also amends the Orphan Drug Act to authorize appropriations of $25 million for FY 2002 for grants and contracts for the development of drugs for rare diseases and conditions.

It defines rare disease as any disease or condition affecting less than 200,000 persons in the United States. More than 300 structural or metabolic birth defects may be included in this definition.

TEXAS 77TH LEGISLATURE: After the May adjournment of the 77th Texas Legislative Session, Governor Rick Perry signed into law over 1500 bills and resolutions. Of these, the following are relevant to those interested in prenatal care, service for chronically ill and disabled children, and medical records privacy.

H.B. 391: Establishes minimum guidelines for the procurement, processing, distribution, or use of human milk by donor milk banks.

H.B. 456: Removes licensing requirement for persons delivering certain personal care and medical care services under a Medicaid or non-Medicaid voucher program to persons with functional disabilities.

H.B. 757: Establishes a nine-member Health Disparities Task Force to be supported by the Texas Department of Health.

H.B. 835: Requires a feasibility study of a family buy-in option for Children's Health Insurance Program (CHIP) with a report due on November 1, 2002.

H.B. 1478: Renames "Work Group on Children's Long-term Care and Health Programs" to "Children's Policy Council (CPC)." Requires CPC to assist state health and human services agencies in developing, implementing, and administering family support policies and related long-term care and health programs for children.

H.B. 3038: Provides for health insurance premium payment for kids who are eligible for Children's Health Insurance Program (CHIP) and Medicaid, if enrollment in a group health insurance plan would be more cost effective.

H.B. 3572: Requires the Health and Human Services Commission

(HHSC), subject to available funds, to award a grant of start-up money to establish an umbilical cord blood bank for recipients of blood and blood components who are unrelated to the donors of the blood.

S.B. 12: Expands the definitions of "genetic information" and "genetic test" in the Texas Labor Code to prevent employers, licensing authorities, and insurance companies from discriminating on the basis of certain genetic information or genetic tests. Prevents employers from discriminating on the basis of family health information, which may contain details that could be used to determine an individual's genetic predisposition to certain diseases.

H.B. 361: Modifies existing legislation relating to the membership and activities of the Interagency Council on Autism and Pervasive Developmental Disorders. Increases consumer membership by adding five consumer positions and allows for reimbursement of public members for travel and related expenses.

H.B. 368: Requires health and human services agencies to develop a permanency plan for institutionalized children; coordinate permanency planning procedures; ensure a child is placed on a waiting list for waiver program services within three days of receiving notice of a child's institutionalization; oversee any permanency planning contractor; and implement an alternative system of care for children who cannot reside with the child's birth family.

H.B. 665: Creates the Office of Early Childhood Coordination (ECC) within the Health and Human Services Commission (HHSC).

READING LIST

- Birth Defects and Infant Mortality: Using data from the National Center for Health Statistics, researchers reported that infant mortality from birth defects declined during 1970-1997, although the decline was slower for nonwhites than for whites. Moreover, the decline was not as steep as for overall infant mortality, so birth defects have become the leading cause of infant mortality. [Obstet Gynecol 2001;98:620-627]
- Birth Defects and Mortality: Investigators in Texas investigated the first-year mortality for 23 birth defect groups. Mortality rates varied by defect, being highest for anencephaly, trisomy 13, and trisomy 18 and lowest for gastroschisis, trisomy 21, and cleft lip with or without cleft palate. [Teratology 2001;64:267-275]
- Elective Termination, Infant Mortality, and Birth Defects: This study compared the impact of elective termination on mortality of infants with birth defects between four European countries. Countries with a higher proportion of elective terminations had lower numbers of fetal and infant deaths associated with birth defects. [Fetal Diagn Ther 2001;16:153-157]
- Birth Defects and Developmental Disabilities: Researchers in Atlanta found increased risk of developmental disabilities with the presence of major birth defects. The association was stronger with increasing severity of the developmental disability. [Pediatrics 2001;108:728-734]
- Neural Tube Defects and Folic Acid Antagonists: Investigators in Boston reported increased risk of neural tube defects with maternal exposure to any folic acid antagonist and the specific antagonists carbamazepine and trimethoprim. Neural tube defect risk in relation to folic acid antagonists was reduced in the presence of folic acid supplements. [Am J Epidemiol;2001:153:961-968]
- Neural Tube Defects and Folic Acid: This randomized double-blind placebo-controlled study in India found periconceptional folic acid supplementation reduced risk of recurrence of neural tube defects. [Indian J Med Res 2000;112:206-211]
- Neural Tube Defects and MTHFR Genotype: Researchers in Mexico reported higher percentages of folate deficiency and the methylenetetrahydrofolate reductase (MTHFR) gene homozygous 677T mutation among mothers of infants and fetuses with neural tube defects than among mothers of controls.[Arch Med Res 2001;32:277-282]
- Spina Bifida and Survival: Researchers in Atlanta found the one year survival rate for spina bifida to be 87.2%. Low birth weight and high location of the lesion were associated with increased mortality. [Paediatr Perinat Epidemiol 2001;15:374-378]
- Congenital Heart Defects and Mortality: Using US death certificate information, investigators found that mortality due to congenital heart defects declined 39% between 1979 and 1997. The decline was identified among all age groups. Moreover, the age at death from congenital heart defects increased over the time period of the study. [Circulation 2001;103:2376-2381]

- Maternal Weight and Heart Defects: Researchers in Atlanta investigated the relationship between maternal prepregnancy weight and infants with heart defects. Underweight women were less likely to have infants with heart defects while overweight women were more likely to have infants with heart defects. Periconceptional multivitamin use reduced risk of having infants with heart defects among both underweight and average weight women, but not among overweight women. [Epidemiology 2001;11:439-446]
- Homocysteine and MTHFR and Cardiac Defects: A study in Alabama found that women with fetuses with cardiac defects had higher amniotic fluid homocysteine levels. Fetuses with cardiac defects were also more likely to have the C677T MTHFR mutation. [Am J Obstet Gynecol 2001;184:806-817]
- Febrile Illness and Heart Defects: Researchers in Atlanta reported increased risk of heart defects with maternal febrile illness during early pregnancy. Increased risk was observed for the specific heart defects of tricuspid atresia, left obstructive defects, transposition of the great arteries, and ventricular septal defects. [Epidemiology 2001;12:485-490]
- Vitamin A and Heart Defects: Using data from the Baltimore-Washington Infant Study, investigators identified increased risk of transposition of the great arteries with maternal supplementation of 10,000 IU or more of retinol. [Epidemiology 2001;12:491-496]
- Oral Clefts and Genes: Researchers in Denmark reported an association between cleft palate and variation in the Transforming Growth Factor-Beta 3 (TGF-B3) gene loci and a potential association between cleft palate and variation in the MSX1 gene loci. No interaction between these loci and maternal cigarette smoking or alcohol use during the first trimester was observed. [Am J Epidemiol 2001;153:1007-15]
- Oral Clefts and Genetic and Environmental Factors: Using data on infants born in Maryland, researchers found no association between oral clefts and maternal smoking, vitamin use, urinary tract infection, and illicit drug use. An association between allele frequencies and oral clefts was found for the MSX1 gene but not for the TGF-alpha, TGF-B3, or BCL3 genes. [Ann Epidemiol 2001;11:434-442]
- Down Syndrome and Mortality: The CDC reported that the median age at death of persons with Down syndrome increased for all racial groups during 1968-1997. However, the median age at death of persons with Down syndrome in 1997 was 50 years for whites, 25 years for blacks, and 11 years for other racial groups. [MMWR 2001;50:463-465]
- Triploidy and Maternal Serum Screening: Researchers in Connecticut found second trimester maternal serum screens of triploidy-affected pregnancies to have either elevated alpha-fetoprotein, elevated human choroinic gonadotropin, and low/normal estriol or low/normal alpha-fetoprotein, low human chorionic gonadotropin, and low estriol. [Prenat Diagn 2001;21:680-686]
- Genitourinary Malformations and Prenatal Diagnosis: An investigation at a major referral center in Boston reported the elective termination rate to be 65% for spina bifida, 46% for posterior urethral valves, 31% for prune belly syndrome, and 25% for bladder exstrophy. [J Urol 2001;165:1677-1680]
- Ear Defects and Renal Defects: Researchers in California reported that 29% of patients with ear defects also had renal defects, and that 92% of those patients with both ear and renal defects were diagnosed with a syndrome involving multiple congenital anomalies. [Pediatrics 2001;108:e32]
- Pyloric Stenosis and Erythromycin: Investigators in Indiana observed an increased risk of hypertrophic pyloric stenosis with systemic erythromycin use in infants, particularly within the first two weeks after delivery. [J Pediatr 2001;139:380-384]
- Birth Defects and Landfills: Researchers in Great Britain found a slightly increased risk of all births defects and of neural tube defects, hypospadias and epispadias, and abdominal wall defects for residents near landfill sites. [BMJ 2001;323:363-368]
- Birth Defects and Trihalomethanes: Researchers in Nova Scotia, Canada, examined the potential relationship between selected birth defects and maternal exposure to chloroform or bromodichloromethane in drinking water. The study found increased risk of neural tube defects with bromodichloromethane, decreased risk of heart defects with bromodichloromethane, a possible increased risk of chromosomal abnormalities with chloroform, and no association between cleft defects and either trihalomethane. [Occup Environ Med 2001;58:443-446]
- Birth Defects and Gulf War Veterans: This study used a survey to obtain information on pregnancy outcome from Gulf War veterans and non-Gulf War veterans. Gulf War veterans were more likely to report birth defects among their offspring. [AnnEpidemiol 2001;11:504-511]

The *Monitor* is published twice a year by the Texas Birth Defects Monitoring Division, Texas Department of Health.

TO BE ADDED TO THE MAILING LIST: For this or other free publications from the Division, please contact us at (512) 458-7232 or e-mail bobbie.mankowski@ tdh.state.tx.us.

MORE INFORMATION CAN BE FOUND AT: www.tdh.state.tx.us/tbdmd/index.htm

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PREGNANCY OUTCOME PATTERNS FOR VARIOUS BIRTH DEFECTS

A SPECIAL REPORT FROM THE TEXAS BIRTH DEFECTS REGISTRY

Birth defects differ in the distribution of pregnancy outcomes of the detected cases. We used Texas birth defects registry data for 1996 and 1997 combined to examine pregnancy outcomes for 50 birth defects. For 1996 and 1997 deliveries, the pregnancy outcomes monitored by the registry included live births of any gestation, spontaneous fetal deaths of at least 20 weeks gestation or 500 grams birth weight, and elective terminations of any gestation. Results of this analysis are shown in the table, and pie charts are shown for selected conditions.

Many birth defects were found to result almost exclusively in live births. For example, 100% of the cases of pyloric stenosis and 99.7% of the cases of atrial septal defect were live births (see Table). Live births made up 95% or more of the total cases for 28 of the 50 conditions examined.

Some birth defects result in many spontaneous fetal deaths at 20 or more weeks of gestation. Examples include anencephaly (26.2% of cases were fetal deaths); agenesis, aplasia, or hypoplasia of the lung (12.4% fetal deaths); and renal agenesis or dysgenesis (10.4% fetal deaths).

Certain conditions frequently result in elective pregnancy terminations. The conditions with a high occurrence of elective terminations are those which are amenable to prenatal diagnosis and which have poor prognoses for survival. Elective terminations made up 43.0% of all anencephaly cases detected; 20.6% of total anencephaly cases were terminations at 20 or more weeks of gestation and 22.4% were terminations before 20 weeks.

Other conditions frequently found among elective terminations include encephalocele (14.3% of all cases were elective terminations at 20+ weeks and 17.1% were terminations before 20 weeks); Patau syndrome (15.6% elective terminations at 20+ weeks and 15.6% terminations before 20 weeks); Edwards syndrome (18.2% elective terminations at 20+ weeks and 10.2% terminations before 20 weeks); and omphalocele (14.7% elective terminations at 20+ weeks and 13.2% terminations before 20 weeks).

NEURAL TUBE DEFECTS

Neural tube defects (NTDs) are a group of birth defects presumed to have a common origin in failure of the neural tube to develop properly during the embryonic stage. NTDs are comprised mainly of anencephaly (including craniorachischisis), spina bifida (including meningomyelocele, meningocele, and myelocele), and encephalocele. Anencephaly involves absence of the skull, with the cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Spina bifida is defective closure of the bony encasement of the spinal cord, through which the cord and meninges may or may not protrude. Encephalocele is protrusion of some or all of the brain through a defect in the skull.

Fig. 1: Anencephaly

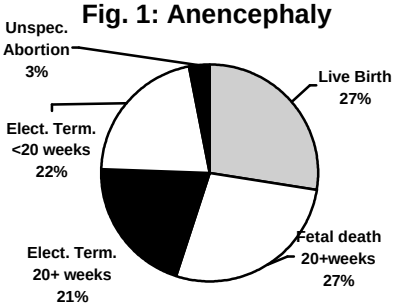
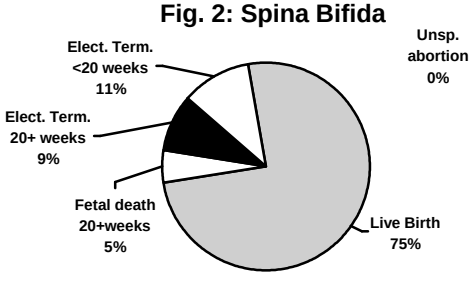
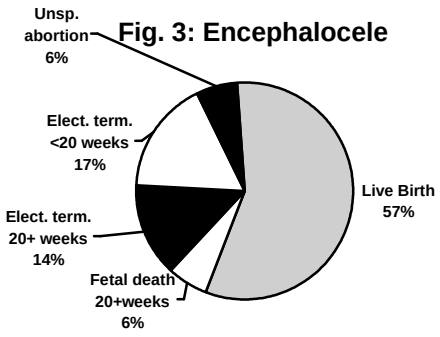


Fig. 2: Spina Bifida



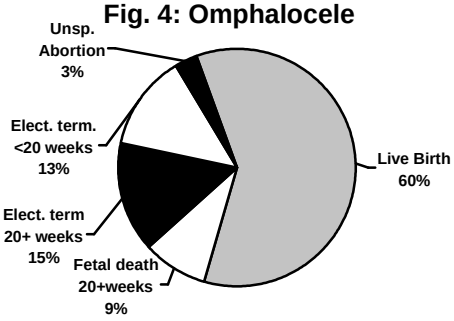


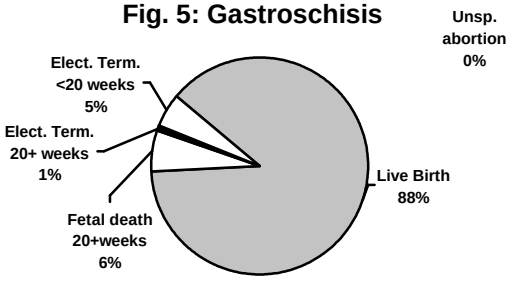
Pregnancy outcome patterns for the various NTDs differ. Among the three NTDs, anencephaly is the least likely to result in a live birth, and the most likely to result in a fetal death or an elective termination. Spina bifida is the most likely of the NTDs to result in a live birth, and the least likely to result in a fetal death or elective termination. The distribution of pregnancy outcomes for encephalocele is in between those for anencephaly and for spina bifida.

ABDOMINAL WALL DEFECTS

Omphalocele is an abdominal wall defect involving herniation of the bowel and liver into the umbilical cord. The defect is always covered by a membrane, although sometimes this membrane may rupture, in which case the defect may be mistaken for gastroschisis. Infants with omphalocele tend to have more additional birth defects, including chromosomal abnormalities (most often trisomy 13, trisomy 18, and trisomy 21), and a lower survival rate than infants with gastroschisis.

Gastroschisis is an abdominal wall defect involving herniation of the intestines and sometimes the liver outside of the abdomen. The defect occurs lateral to the umbilicus, usually on the right, and never includes a covering sac. The defect may be confused with other abdominal wall defects such as omphalocele and body stalk anomaly. Infants with gastroschisis tend to have fewer additional birth defects, including chromosomal abnormalities, and a higher survival rate than infants with other abdominal wall defects, particularly omphalocele.





The pregnancy outcome patterns for omphalocele and gastroschisis are very different. Omphalocele is much less likely to result in a live birth, and much more likely to result in a fetal death or elective termination, than gastroschisis.

TRISOMIES

Down syndrome (trisomy 21) is the most common autosomal abnormality among live births, and is caused by an extra copy of chromosome 21. Down syndrome is associated with a variety of structural malformations, particularly cardiac malformations. Patau syndrome (trisomy 13), caused by an extra copy of chromosome 13, is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants with Patau syndrome do not survive beyond 6 months of life. Edwards syndrome (trisomy 18), caused by an extra copy of chromosome 18, is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits; cardiac and renal anomalies are also common. For infants with Edwards syndrome, survival of more than a few months is rare.

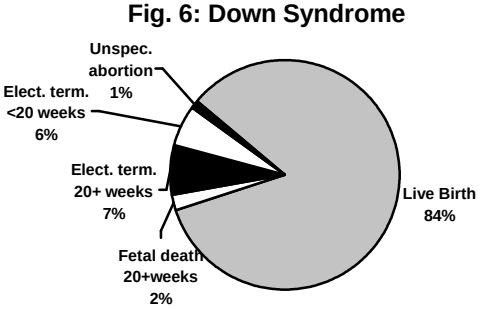


Fig. 7: Edwards Syndrome

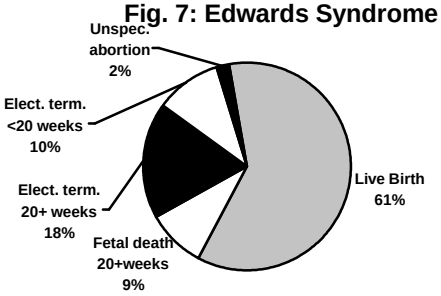
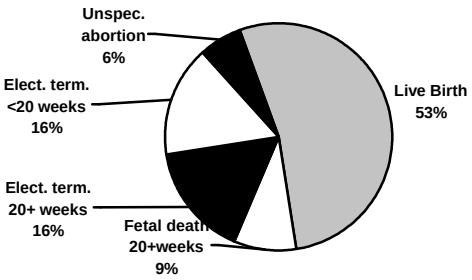


Fig. 8: Patau Syndrome



The patterns of pregnancy outcomes for these three trisomies differ from each other. Among the three trisomies, Down syndrome is the most likely to result in a live birth, and the least likely to result in a fetal death or an elective termination. Patau syndrome is the least likely of the trisomies to result in a live birth, and the most likely to result in a fetal death or elective termination. The distribution of pregnancy outcomes for Edwards syndrome is in between those for Down syndrome and for Patau syndrome.

OTHER SE ECTED DEFECTS

Renal agenesis or dysgenesis includes the absence of one or both of the kidneys or a deviation in the embryonic development of the kidney. Unilateral renal agenesis may be asymptomatic and is often incidentally diagnosed by abdominal ultrasound or computed tomography (CT) scan secondary to another condition. Bilateral renal agenesis is invariably fatal. Agenesis, aplasia, or hypoplasia of the lung includes the absence or incomplete development of the lung or lung tissue. When bilateral renal agenesis occurs, hypoplasia of the lung is almost always a co-occurring condition.

Examination of these two groups revealed that 23.6% of the cases with a diagnosis in the category renal agenesis/dysgenesis also had agenesis, aplasia, or hypoplasia of the lung, and 24.8% of the cases with agenesis, aplasia, or hypoplasia of the lung also had renal agenesis/dysgenesis. While there is a fair amount of overlap between these two groups, the co-occurrence of these defects cannot completely account for the striking similarity in pregnancy outcome patterns for these two defects.

Fig. 9: Renal Agenesis/Dysgenesis

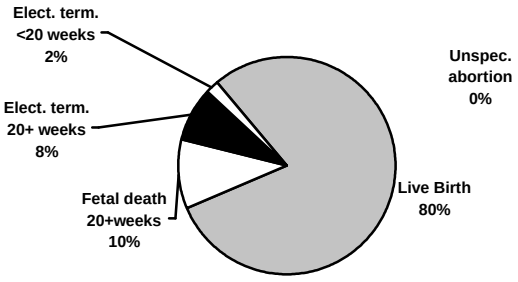
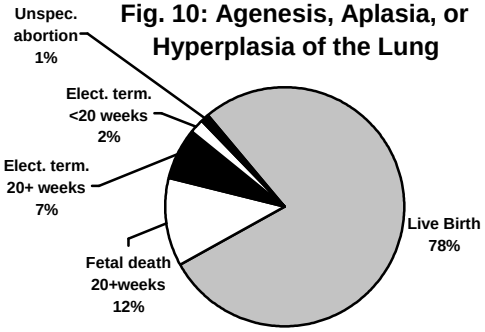


Fig. 10: Agenesis, Aplasia, or Hyperplasia of the Lung



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Pregnancy outcome patterns for various defects, Texas, 1996 and 1997 deliveries.

Defect	Live Birth		Spontaneous fetal death at 20+ weeks of gestation		Induced termination at 20+ weeks of gestation		Induced termination < 20 weeks of gestation		Unspecified abortion (termination vs. fetal death)		Total	
	number	row %	number	row %	number	row %	number	row %	number	row %	number	row %
Anencephaly	30	28.0%	28	26.2%	22	20.6%	24	22.4%	3	2.8%	107	100.0%
Spina bifida without anencephaly	114	76.0%	7	4.7%	13	8.7%	16	10.7%			150	100.0%
Encephalocele	20	57.1%	2	5.7%	5	14.3%	6	17.1%	2	5.7%	35	100.0%
Microcephaly	166	97.1%	5	2.9%							171	100.0%
Holoprosencephaly	32	69.6%	6	13.0%	6	13.0%	1	2.2%	1	2.2%	46	100.0%
Hydrocephaly	191	84.5%	8	3.5%	18	8.0%	5	2.2%	4	1.8%	226	100.0%
Anophthalmia	11	68.8%	2	12.5%	2	12.5%	1	6.3%			16	100.0%
Microphthalmia	69	98.6%			1	1.4%					70	100.0%
Cataract	31	100.0%									31	100.0%
Aniridia	2	100.0%									2	100.0%
Anotia or microtia	76	90.5%	1	1.2%	6	7.1%			1	1.2%	84	100.0%
Common truncus	18	94.7%			1	5.3%					19	100.0%
Transposition of the great vessels	142	98.6%	1	0.7%	1	0.7%					144	100.0%
Tetralogy of Fallot	83	95.4%	2	2.3%	2	2.3%					87	100.0%
Ventricular septal defect	1284	97.9%	12	0.9%	12	0.9%	3	0.2%	1	0.1%	1312	100.0%
Atrial septal defect	1419	99.7%	2	0.1%	2	0.1%					1423	100.0%
Endocardial cushion defect	95	92.2%	2	1.9%	2	1.9%	3	2.9%	1	1.0%	103	100.0%
Pulmonary valve atresia or stenosis	143	98.6%	1	0.7%	1	0.7%					145	100.0%
Tricuspid valve atresia or stenosis	69	95.8%			3	4.2%					72	100.0%
Ebstein anomaly	11	84.6%	1	7.7%					1	7.7%	13	100.0%
Aortic valve stenosis	68	100.0%									68	100.0%
Hypoplastic left heart syndrome	69	97.2%			1	1.4%	1	1.4%			71	100.0%
Patent ductus arteriosus	1624	99.8%	3	0.2%					1	0.1%	1628	100.0%
Coarctation of the aorta	142	97.9%			2	1.4%			1	0.7%	145	100.0%
Choanal atresia or stenosis	34	94.4%	1	2.8%	1	2.8%					36	100.0%
Agenesis, aplasia, or hypoplasia of the	107	78.1%	17	12.4%	9	6.6%	3	2.2%	1	0.7%	137	100.0%
Cleft palate alone (without cleft lip)	177	98.3%	1	0.6%	2	1.1%					180	100.0%
Cleft lip with or without cleft palate	316	90.3%	15	4.3%	14	4.0%	4	1.1%	1	0.3%	350	100.0%
Tracheoesophageal fistula / esophageal	69	98.6%	1	1.4%							70	100.0%
Pyloric stenosis	482	100.0%									482	100.0%
Stenosis or atresia of small intestine	93	96.9%	1	1.0%			1	1.0%	1	1.0%	96	100.0%
Stenosis or atresia of large intestine, rectum, or anal canal	112	88.9%	8	6.3%	3	2.4%	3	2.4%			126	100.0%
Hirschsprung disease	39	100.0%									39	100.0%
Biliary atresia	17	100.0%									17	100.0%
Hypospadias or epispadias	718	99.7%	1	0.1%	1	0.1%					720	100.0%
Renal agenesis or dysgenesis	114	79.2%	15	10.4%	12	8.3%	3	2.1%			144	100.0%
Obstructive genitourinary defect	493	97.8%	3	0.6%	6	1.2%	2	0.4%			504	100.0%
Bladder exstrophy	5	100.0%									5	100.0%
Congenital hip dislocation	168	99.4%			1	0.6%					169	100.0%
Reduction defects of the upper limbs	112	88.2%	8	6.3%	3	2.4%	4	3.1%			127	100.0%
Reduction defects of the lower limbs	33	82.5%	3	7.5%	3	7.5%	1	2.5%			40	100.0%
Craniosynostosis	79	97.5%	1	1.2%	1	1.2%					81	100.0%
Diaphragmatic hernia	59	89.4%	4	6.1%	3	4.5%					66	100.0%
Omphalocele	41	60.3%	6	8.8%	10	14.7%	9	13.2%	2	2.9%	68	100.0%
Gastroschisis	94	88.7%	6	5.7%	1	0.9%	5	4.7%			106	100.0%
Down syndrome	316	84.0%	9	2.4%	27	7.2%	21	5.6%	3	0.8%	376	100.0%
Patau syndrome	17	53.1%	3	9.4%	5	15.6%	5	15.6%	2	6.3%	32	100.0%
Edwards syndrome	53	60.2%	8	9.1%	16	18.2%	9	10.2%	2	2.3%	88	100.0%
FAS or other alcohol related birth defects	2	100.0%								100.0%		
Possible/probable FAS or other alcohol related birth defects	18	100.0%									18	100.0%
TOTAL	9677	94.4%	194	1.9%	218	2.1%	130	1.3%	28	0.3%	10247	100.0%